

Patient Informed Consent Form for Genetic Testing

I _____ (Patient's Name), authorize Athena Diagnostics to conduct genetic testing for _____ (Disease and/or Test Name), as ordered by my physician or authorized healthcare provider or my child's or dependent's physician or authorized healthcare provider. In the following paragraphs, "you" or "your" may refer to you or your child/dependent. If the person getting the tests performed lacks the capacity to consent, this consent form will need to be signed by a person authorized to consent for that person. Please understand that due to the complexity of genetic testing and the important implications of the test results, these results will be reported only to the patient's ordering physician or designee and that you must contact your physician to obtain the results of the tests. Please read this information and feel free to ask your physician questions before you agree to get tested. Some words are starred when they are first used. The definitions are in alphabetical order in the **Glossary** on the last page.

What is Involved?

A blood, body fluid, or tissue specimen will be collected from you. This specimen will be treated to isolate DNA*, RNA* or gene products* for genetic testing or chemical analysis. The test will cover all disorders requested by your physician on the Athena Diagnostics requisition form.

A Why is the Genetic Test(s) Being Done?

The purpose of genetic testing is to look for changes in genes* or gene products that cause a disease. Such testing may include DNA, RNA or their products. The test may help determine whether you are affected with, or at risk to someday be affected with, a specific genetic disorder.

What is a Mutation?

Mutations* are errors in these instructions that could cause a protein not to be produced or not work properly. In these ways, mutations may cause a disease, or make someone more likely to develop a disease. Your physician has decided that you may have a mutation* or change in your genetic code* or you have changes in gene products that have led to a genetic disease. These changes may be due to a loss (deletion) gain (duplication), or substitution of genetic material. The changes may include one or more parts of a gene, an entire gene or multiple genes. These changes may also be found in your family members who may also have the same changes. The testing described above is being ordered by your physician to see if you have change(s) in genes or gene products, as limited by the test(s) ordered.

What are the Ways to Find Mutations?

One method to find changes in DNA or RNA is called sequencing. New methods called whole exome* (WES) and whole genome* sequencing (WGS) provide a way to analyze all or some of your gene sequences in one blood draw. These methods will allow the testing laboratory to determine an important piece of your genetic code. Your physician will evaluate this information in relation to your medical condition or the medical condition of your family member(s). Genetic testing, in general, and new methods such as WES/WGS, in particular, does not identify all types of genetic changes such as losses, duplications, or substitutions of genetic material. Genetic testing may identify changes that place you at risk to develop a disease in the future or may reveal changes that have unknown significance.

What are the Physical Risks?

Most genetic tests are blood tests. Blood tests involve collection of a small amount of blood using routine collection procedures that carry minimal risk. You may experience some discomfort at the site of needle entry, and there is a risk of a "black and blue" mark. There is a remote risk of fainting or local infection.



What are Other Potential Risks?

Genetic testing can reveal many things about you, including the mutations that may cause a disease in you or members of your family. Some of the information may be important to your present or future health, some of it may have nothing to do with your health and for much of it we will not know how it might or might not affect you or your family's health. There is also the potential that genetic analysis will reveal unwanted information, for example about ancestry, parentage, other non-medical physical or personality traits, etc. The data may also reveal that you may be at risk for certain genetic diseases or if you are a carrier of disease-associated mutations. Also, predictions about health and disease from sequencing are not 100 percent accurate. The Genetic Information Nondiscrimination Act (GINA) generally protects you against discrimination based on your genetic information when it comes to health insurance and employment.

What if the Results are Uncertain?

You may learn that a variant of uncertain clinical significance was identified by a genetic test. This means that a genetic change (variant) was identified, but it is unknown whether the variant is the cause of medical problems in you. The variant could be a benign change that does not cause medical problems or it could be a disease-causing abnormality. Parental testing may be recommended to find out if a parent also carries the genetic variant or to determine the variant's clinical significance.

What Do the Test Results Mean?

The test results may indicate that you have a genetic disease or are at increased risk to be affected at a later age, or at-risk to have future affected children with this condition. It is important for you to understand that there is a possibility that the test results may not be able to determine whether you have a genetic disease or will develop one in the future.

If the results of the molecular genetics tests are positive, that means that an individual may be a carrier of a certain disease, be predisposed to developing a certain disease or actually have the specific disease or condition that was ordered by your physician. You may want to consider further independent testing, consult with your physician or pursue genetic counseling even if the results are negative.

It is important to understand the limitations of testing and to discuss these limitations with your physician or genetic counselor before testing. For example, the interpretation of test results could be based upon probabilities, and may not provide a 100 percent definitive answer to whether or not you have a specific genetic disease or are at-risk for developing the disease.

What Should Happen Before and After Testing?

Your physician or other authorized person, such as a genetic counselor, should provide you with the following:

1. A discussion of the purpose and description of the test and a general written description of each specific disease or condition tested for.
2. Pre-test counseling regarding the expected outcomes of testing, the likelihood and type of incidental results that could be generated and what results will or will not be disclosed. These test results can include gene variants not known to be specifically associated with a disease or gene variants known to be associated with a disease but not believed to be related to the condition that led to the testing ("secondary findings"). Secondary findings will not be reported unless specifically ordered by your physician after consultation with a medical geneticist and a genetic counselor.
3. Information regarding the reliability of positive or negative test results, and the level of certainty that a positive test result for that disease or condition serves as a predictor of such disease.
4. Post-test counseling to provide information about appropriate specialist interventions associated with clinically relevant results.

Will Your Specimen or Data Be Used for Other Purposes?

Genetic specimens may be kept by the laboratory for additional diagnostic testing as indicated above and may be used for internal laboratory quality assurance purposes. Genetic specimens can only be retained for 60 days for New York State residents.

Athena Diagnostics requests your permission to use your specimen, clinical information and data in a de-identified way (without any identifying information) for research, educational studies, commercial purposes and/or publication, if appropriate. Your name or other personal identifying information will not be used in or linked to the results of any studies and publications. Any specimens that you have donated which are used in research may result in new products, tests or discoveries. In some instances, this may have potential commercial value and may be developed and owned by the Athena Diagnostics and/or others. However, donors do not retain any property rights to the materials. Therefore, you would not share in any financial benefits from these products, tests or discoveries. If you change your mind and do want to use your specimen for research, it will not be possible to reliably retrieve your specimen or data once they are de-identified. Your refusal to have your specimen used for research purposes will not affect your results. Please indicate your approval or denial on the Athena Test Requisition Form under *Authorization to Use De-Identified Specimen or Data for Research*.

What About Confidentiality?

Results will only be released to the ordering physician or designee and/or others as entitled by law. As described above, information obtained from the test may be used in scientific publications or presentations, but the identity of all individuals studied will not be revealed in such publications or presentations.

Health Provider Statement

By their signature below, the healthcare provider indicates that he or she has explained the purpose of the test, the procedures, the benefits and risks that are involved in testing to their patient. His or her patient has been given the opportunity to ask questions about this consent and seek genetic counseling. The health care provider acknowledges that his or her patient has voluntarily decided to have the test performed at Athena Diagnostics.

Signature of Person Obtaining Consent

Date

Printed Name of Person Obtaining Consent

Patient's Statement

I, the undersigned, have been informed about the test(s) purpose, procedures, possible benefits and risks, and I have received a copy of this consent. I have been given the opportunity to ask questions before I sign, and I have been told that I can ask other questions at any time. I voluntarily agree to genetic testing.

Signature of Patient

Date

Printed Name of Patient

Signature of Parent or Legally Authorized Representative

Date

Printed Name of Parent or Legally Authorized Representative

Relationship to Participant



Glossary

DNA (deoxyribonucleic acid): The molecule that encodes genetic information. DNA is a double- stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases: adenine (A), guanine (G), cytosine (C) and thymine (T). In nature, base pairs form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner.

Chromosomes: The self replicating genetic structures of cells containing the cellular DNA that bears in its nucleotide sequence the linear array of genes.

Exome: All the genetic material in the parts of the chromosomes of a human that code for proteins.

Gene: The fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product (i.e., a protein or RNA molecule).

Genetic code: The sequence of nucleotides, coded in triplets (codons) along the mRNA that determines the sequence of amino acids in protein synthesis. The DNA sequence of a gene can be used to predict the mRNA sequence, and the genetic code can in turn be used to predict the amino acid sequence.

Gene product: The biochemical material, either RNA or protein, resulting from expression of a gene. The amount of gene product is used to measure how active a gene is; abnormal amounts can be correlated with disease causing alleles.

Genome: All the genetic material in the chromosomes of a human.

Mutation: Any heritable change in DNA sequence.

Protein: A large molecule composed of one or more chains of amino acids in a specific order; the order is determined by the base sequence of nucleotides in the gene coding for the protein. Proteins are required for the structure, function and regulation of the body's cells, tissues and organs, and each protein has unique functions. Examples are hormones, enzymes and antibodies.

RNA (ribonucleic acid): A chemical found in the nucleus and cytoplasm of cells; it plays an important role in protein synthesis and other chemical activities of the cell. The structure of RNA is similar to that of DNA. There are several classes of RNA molecules, including messenger RNA, transfer RNA, ribosomal RNA and other small RNAs, each serving a different purpose.



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